

Sport Preparticipation Screening for Asymptomatic Atlantoaxial Instability in Patients With Down Syndrome

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Abstract: Down syndrome (DS) is a clinical syndrome comprising typical facial features and various physical and intellectual disabilities due to extra genetic material on chromosome 21, with one in every 1000 babies born in the United Kingdom affected. Patients with Down syndrome are at risk of atlantoaxial instability (AAI). Although AAI can occur in other conditions, such as rheumatoid arthritis, this position statement deals specifically with patients with DS and asymptomatic AAI. Atlantoaxial instability, also referred to as atlantoaxial subluxation, is defined as increased movement between the first (atlas) and second (axial) cervical vertebra joint articulation, the atlantoaxial joint. Atlantoaxial instability is reported to occur in 6.8% to 27% of the DS population, although this varies depending on the age of the patients whom you are screening. Less than 1% to 2% of these patients are then thought to later develop symptomatic AAI, although the natural history and progression of AAI is not well understood. The risks associated with AAI are neurological injury from excessive movement of the cervical vertebra impinging on and then damaging the spinal cord, although the risk of this during sporting activities is extremely rare. Clearly, physical activity and sports participation for patients with DS has many biological, psychological, and social benefits, and the Faculty of Sport and Exercise Medicine (FSEM), United Kingdom, wishes to promote safe physical activity and sport for all. The FSEM, United Kingdom, has therefore produced a statement regarding sport preparticipation screening for asymptomatic AAI in patients with DS.

Key Words: down syndrome, sport preparticipation screening, atlantoaxial instability, atlantoaxial subluxation

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INTRODUCTION

Down syndrome (DS) is a clinical syndrome comprising typical facial features and various physical and intellectual disabilities due to extra genetic material on chromosome 21, with one in every 1000 babies born in the United Kingdom affected.¹ Patients with Down syndrome are at risk of atlantoaxial instability (AAI). Although AAI can occur in other conditions, such as rheumatoid arthritis, this position statement deals specifically with patients with DS and asymptomatic AAI.

Atlantoaxial instability, also referred to as atlantoaxial subluxation, is defined as increased movement between the first (atlas) and second (axial) cervical vertebra joint articulation, the

atlantoaxial joint.² Atlantoaxial instability in patients with DS is due to a combination of ligament laxity and bony abnormalities of the atlantoaxial joint. Atlantoaxial instability is reported to occur in 6.8% to 27% of the DS population,^{3,4} although this varies depending on the age of the patients whom you are screening. Less than 1% to 2%^{3,5} of these patients are then thought to later develop symptomatic AAI, although the natural history and progression of AAI is not well understood.²

The risks associated with AAI are neurological injury from excessive movement of the cervical vertebra impinging on and then damaging the spinal cord. The risk of this during sporting activities is extremely rare.^{2,6} Clearly, physical activity and sports participation for patients with DS has many biological, psychological, and social benefits.⁷ The Faculty of Sport and Exercise Medicine (FSEM), United Kingdom, wishes to promote safe physical activity and sport for all. Patients with Down syndrome are at risk of asymptomatic AAI, and the FSEM, United Kingdom, has therefore produced a position statement dealing specifically with this concern.

PLAIN LATERAL CERVICAL SPINE RADIOGRAPHY AS A SCREENING TEST FOR ASYMPTOMATIC ATLANTOAXIAL INSTABILITY

1. Cremers et al⁶ followed up 91 patients with asymptomatic AAI for 1 year after screening them with lateral cervical spine radiography, letting them either participate in unrestricted sport or restricting their sporting activities. They found no difference between the groups in terms of motor or neurological function. They concluded that there is no evidence to support plain radiography screening for asymptomatic AAI.

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N. Heron led the conception and design of the position paper and prepared the first draft of the manuscript. All authors were involved in critical revisions and reviewing background reading. All authors critically reviewed the manuscript and approved the final version submitted for publication. All authors read and approved the final manuscript.

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2. There are diagnostic inconsistencies surrounding asymptomatic AAI on plain radiography, with, eg, Morton et al⁸ reporting that it occurs at 4 mm, whereas other authors reporting that it occurs at 3 mm^{9,10} and some at 4.5 mm.⁵
 3. Atlantoaxial instability diagnosis on plain x-ray can also change as the patient gets older.⁴ In 1986, Morton et al⁸ followed up 90 children aged between 4 and 19 years for 5 years after screening them with plain cervical x-ray and, overall, the AAI prevalence decreased at follow-up.
 4. Obtaining plain lateral cervical spine radiography and then the necessary measurements for AAI is technically difficult.⁶
 5. Symptomatic AAI is rare in patients with DS.²
 6. Plain lateral cervical spine radiography (including neutral, flexion, and/or extension views) cannot therefore be recommended as a screening test for asymptomatic AAI in sport.^{2,4,6,8,10}
- Butterfly stroke and diving starts at swimming;
 - Pentathlon;
 - Contact sports such as martial arts, rugby, and soccer; and
 - High jump.
2. Atlantoaxial instability symptoms for patients, family members, and health and sport professionals to be aware of include:^{4,11,13}
 - Change in gait or use of arms or hands,
 - Change in bowel or bladder function,
 - Neck pain,
 - Stiff neck,
 - Head tilt,
 - How the child positions his or her head, and
 - Change in general function, or weakness.
 3. These “warning” symptoms need to be promoted to the general population, patients, family members, and health professionals, eg, through a public health campaign,⁴ to allow them to act if they identify an issue.
 4. Specific signs and symptoms for health professionals to be aware of when they are examining a patient with DS at risk of AAI include:²
 - Easy fatigability;
 - Difficulties in walking;
 - Abnormal gait;
 - Neck pain;
 - Torticollis or head tilt;
 - Incoordination and clumsiness;
 - Sensory deficits;
 - Spasticity;
 - Hyperreflexia;
 - Clonus;
 - Extensor-plantar reflex; and
 - Other upper motor neuron and posterior column signs and symptoms.

Alternative Sport Preparticipation Screening Techniques for Asymptomatic Atlantoaxial Instability

1. For sport preparticipation assessment, a focused history and neurological examination of patients with DS should be undertaken by an appropriately qualified medical professional or chartered physiotherapist,^{4,6} with the preference for the professional to be someone who cares for the patient regularly, on an ongoing basis and is therefore aware of their baseline function,⁶ eg, general practitioner/family physician.
2. Selby et al¹⁰ conclude that both plain x-ray and physical examination are insensitive for screening for AAI in patients with DS.
3. For the neurological examination, Morton et al⁸ advise checking for gait disturbance, neck movements, tendon reflexes, and plantar responses. This is similar to British gymnastics, who have developed their own information sheet and undertake their own screening questions, with no radiological screening.¹¹ This is also supported by Down Syndrome Association.¹²

The 3 screening questions are as follows:¹¹

- A) Does the person show evidence of progressive myopathy? Yes/No
- B) Does the person have poor head/neck muscular control? Yes/No
- C) Does the person's neck flexion allow the chin to rest on their chest? Yes/No

To help with question B, the person's neck control can be assessed by: laying the person on their back with legs straight and they are then pulled to a sitting position by their hands, with the examiner pulling them from the front.¹¹

Depending on the results of the neurological examination, assessment of neck control, and the 3 screening questions, there will essentially be 2 options for the patient:

- Unrestricted sports participation;
- Restricted sports participation

HIGH-RISK SPORTS FOR PATIENTS WITH DOWN SYNDROME AND SYMPTOMS OF ATLANTOAXIAL INSTABILITY TO BE AWARE OF

1. Sports considered to put patients with DS at higher risk of developing symptomatic AAI include:²
 - Gymnastics including trampolining;
 - Diving;

Acute Management of Symptomatic Atlantoaxial Instability

1. If you suspect symptomatic AAI, the patient's spine should be immobilized and an urgent assessment, including neurosurgical consult, should be undertaken within the emergency department along with MRI spinal imaging.⁵

Promoting Safe Sport for Patients With Down Syndrome

1. To facilitate safe sport for patients with DS, we need to promote neck conditioning exercises for this cohort of patients, similar to the neck program promoted in patients with chronic neck discomfort;¹⁴ however, there are no current scientific studies within patients with DS to support this statement. In addition, we need to promote safe sporting practices, eg, appropriate supervision when undertaking higher-risk sports such as trampolining or rugby.

CONCLUSIONS

Plain radiography cannot be currently recommended to screen for asymptomatic AAI. Patients with Down syndrome undergoing a sport preparticipation screening should have 3 questions asked as per the British Gymnastics program and a neurological and neck control assessment should be

undertaken. Neck conditioning exercises should be generally promoted among the DS population. Awareness of potential signs and symptoms of symptomatic AAI need to be raised among patients with DS, family members and professionals caring for this group of patients, as well as the need to provide appropriate supervision when patients with DS are undertaking certain “high-risk” sports. These measures will then allow patients with DS to maximize the biological, social, and psychological benefits of physical activity and sport participation.

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